CASE REPORT

Late gadolinium enhanced cardiovascular magnetic resonance in Becker muscular dystrophy

A Varghese, D J Pennell

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Becker muscular dystrophy is a rare cause of dilated cardiomyopathy. A case of Becker muscular dystrophy is reviewed in which cardiovascular magnetic resonance showed previously unreported findings of extensive mid-myocardial late gadolinium enhancement. Similar detection of late gadolinium enhancement in conjunction with other uses of cardiovascular magnetic resonance may contribute significantly to the diagnosis and management of patients with this unusual and important diagnosis.

A 37 year old man presented with dyspnoea following a flu-like illness. He had a maternal uncle who had died of dilated cardiomyopathy aged 60 years, a maternal grandfather who died of bronchopneumonia and myocardial infarction aged 53 years, and a maternal great grandfather who was wheelchair bound from his 60s and died of endocardial fibrosis and progressive muscular atrophy aged 81. Lateral T wave inversion was noted on 12 lead ECG with a dilated and impaired left ventricle on transthoracic echocardiography. Viral titres were unremarkable. Coronary angiography and right ventricular biopsy were normal. Skeletal muscle biopsy from the left vastus lateralis showed a dystrophic process with subsequent immunostaining for dystrophin II and DAG 43 confirming a deficiency of these proteins and hence a diagnosis of Becker muscular dystrophy (BMD). The patient was treated for symptomatic cardiac failure. Recent deterioration in cardiopulmonary exercise testing led to his consideration for biventricular pacing and cardiovascular magnetic resonance (CMR). CMR showed a thinned and dilated left ventricle with impaired biventricular function, and late gadolinium enhancement showed extensive mid myocardial enhancement anteroseptally (figs 1 and 2).

DISCUSSION

BMD and Duchenne muscular dystrophy are X linked recessive neuromuscular disorders characterised by progressive muscular weakness in the cardiac and skeletal muscle. They are allelic variants of mutations in the gene encoding dystrophin.[1] Death of patients with BMD is more commonly caused by cardiac failure following dilated cardiomyopathy compared with respiratory failure in Duchenne muscular dystrophy.[2] This distinction may partly be because patients with BMD are less limited by skeletal muscle involvement and can perform more strenuous exercise, which is harmful for dystrophin deficient myocardial cells.[3] [4] Cardiac involvement is invariably present in BMD but its severity cannot be predicted by age, degree, and duration of skeletal muscle weakness, type of mutation, or presence of cardiac symptoms, which are usually absent. Cardiac myocyte dystrophin deficiency leads to fibre necrosis causing biventricular replacement of morbid myocardium with connective tissue or fat.[5] The mid wall of the inferolateral left ventricle is most commonly affected, with conduction system disease occurring late.[6] Regular review with ECG and transthoracic echocardiography is recommended.[7] Pharmacological options include β blockers and angiotensin converting enzyme
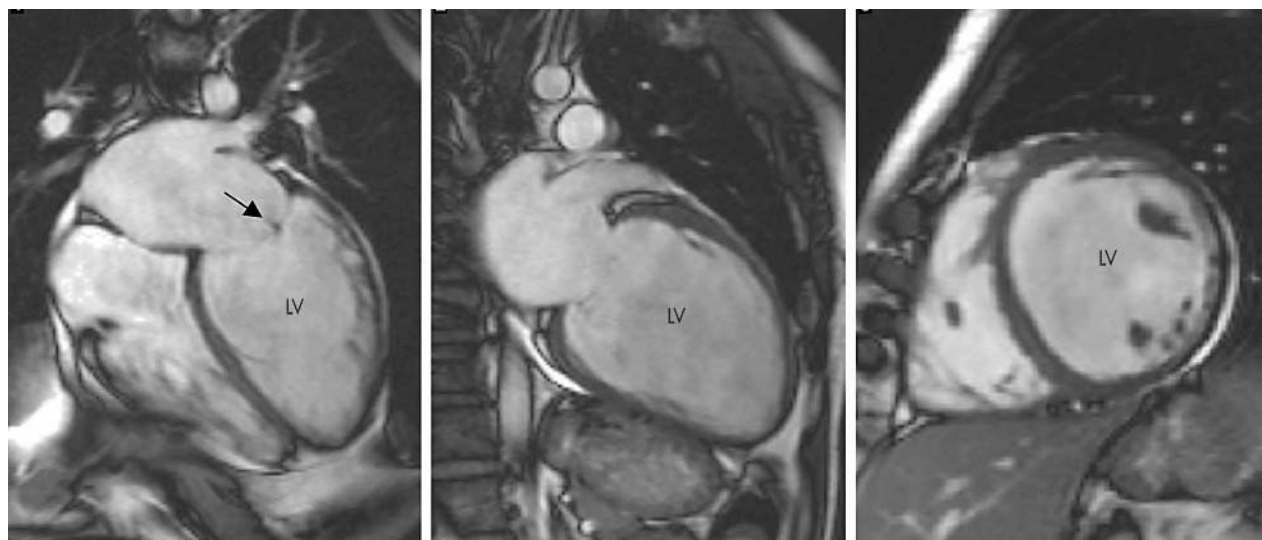

Figure 1 Cine cardiovascular magnetic resonance images in four chamber, vertical long axis, and short axis views. These showed biventricular functional impairment and a dilated left ventricle (LV). Mild functional mitral regurgitation was also noted (arrow).
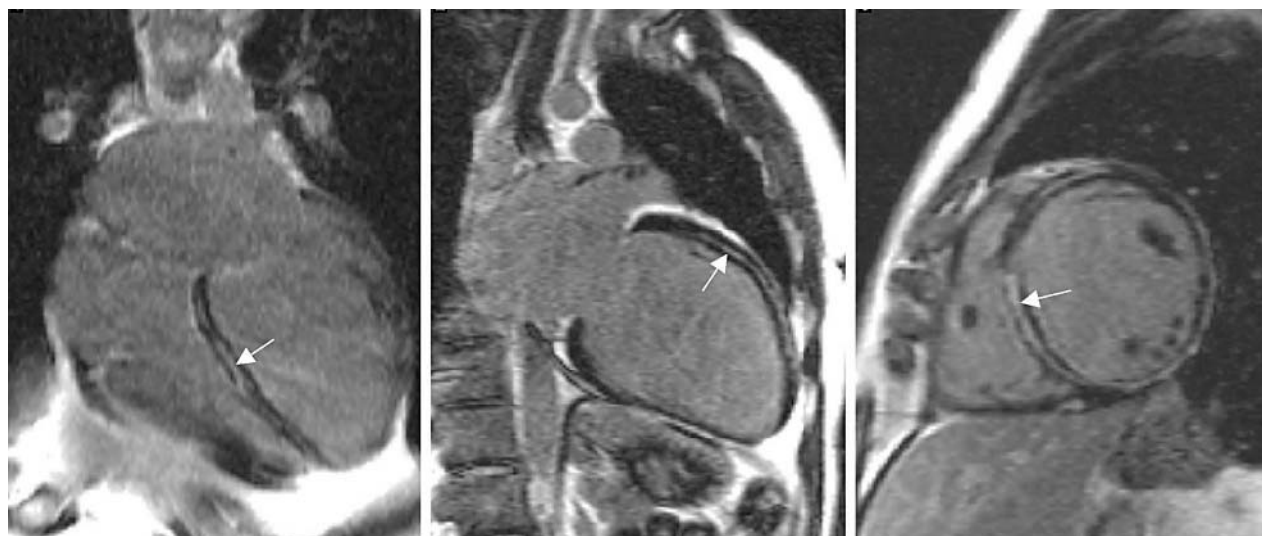
Figure 2 Corresponding images obtained with late gadolinium enhancement. Extensive areas of mid myocardial enhancement are seen within the septum and anterior wall (arrows).

inhibitors; surgical options include cardiac transplantation. Probands are screened at the time of diagnosis whereas carriers need surveillance after 16 years of age.

About 10% of female carriers develop overt cardiac failure. Severity of cardiac involvement increases with time but progression is unpredictable. ECG abnormalities such as pathological Q waves in the inferolateral leads and ST segment and T wave changes are early indicators of left ventricular involvement with left bundle branch block noted in advanced cases.[8] Diffuse hypoperfusion on thallium-201 myocardial single photon emission computed tomography implies severe myocardial dystrophy or fibrosis and correlates with adverse prognosis.[9] Right ventricular involvement has been noted, particularly in the teenage BMD population by transthoracic echocardiography, but this paediatric predilection may reflect the limitations of this assessment with advancing age.

We recently reported mid-myocardial gadolinium enhancement in a dilated cardiomyopathy subgroup, which is thought to correlate with focal segmental fibrosis at autopsy.[10] Gadolinium enhanced CMR distinguishes left ventricular dysfunction related to dilated cardiomyopathy or coronary artery disease on the basis of myocardial patterns of enhancement. In coronary artery disease, a subendocardial or transmural pattern is seen while mid wall changes suggest dilated cardiomyopathy. Mid myocardial gadolinium enhancement has not previously been reported in BMD.

CMR has several potential uses: non-invasive surveillance of probands and screening of carriers without exposure to ionising radiation; prognostication and risk stratification alone or in conjunction with ECG data such as the cardiomyopathic index; determination of necessity and timing of biventricular pacemaker or automatic implantable cardiac defibrillator placement in preference to right ventricular biopsy to assess cardiac fibrosis and fatty change; and preclinical detection of right and left ventricular dilatation to optimise pharmacological intervention and provide lifestyle advice in a timely manner. In addition, mid myocardial gadolinium enhancement in dilated cardiomyopathy should highlight the need to exclude this rare but important diagnosis.

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Authors' affiliations
A Varghese, D J Pennell, Cardiovascular Magnetic Resonance Unit, Royal Brompton and Harefield NHS Trust, London, UK

Correspondence to: Dr A Varghese, Cardiovascular Magnetic Resonance Unit, Royal Brompton Hospital, Sydney Street, London SW3 6NP, UK; A.Varghese@rbh.nthames.nhs.uk

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